glucose-6-phosphate dehydrogenase deficiency

Glucose-6-phosphate dehydrogenase deficiency is a genetic disorder that occurs most often in males. This condition mainly affects red blood cells, which carry oxygen from the lungs to tissues throughout the body. In affected individuals, a defect in an enzyme called glucose-6-phosphate dehydrogenase causes red blood cells to break down prematurely. This destruction of red blood cells is called hemolysis.

The most common medical problem associated with glucose-6-phosphate dehydrogenase deficiency is hemolytic anemia, which occurs when red blood cells are destroyed faster than the body can replace them. This type of anemia leads to paleness, yellowing of the skin and whites of the eyes (jaundice), dark urine, fatigue, shortness of breath, and a rapid heart rate. In people with glucose-6-dehydrogenase deficiency, hemolytic anemia is most often triggered by bacterial or viral infections or by certain drugs (such as some antibiotics and medications used to treat malaria). Hemolytic anemia can also occur after eating fava beans or inhaling pollen from fava plants (a reaction called favism).

Glucose-6-dehydrogenase deficiency is also a significant cause of mild to severe jaundice in newborns. Many people with this disorder, however, never experience any signs or symptoms.

Frequency

An estimated 400 million people worldwide have glucose-6-phosphate dehydrogenase deficiency. This condition occurs most frequently in certain parts of Africa, Asia, and the Mediterranean. It affects about 1 in 10 African American males in the United States.

Genetic Changes

Mutations in the *G6PD* gene cause glucose-6-phosphate dehydrogenase deficiency.

The *G6PD* gene provides instructions for making an enzyme called glucose-6-phosphate dehydrogenase. This enzyme is involved in the normal processing of carbohydrates. It also protects red blood cells from the effects of potentially harmful molecules called reactive oxygen species. Reactive oxygen species are byproducts of normal cellular functions. Chemical reactions involving glucose-6-phosphate dehydrogenase produce compounds that prevent reactive oxygen species from building up to toxic levels within red blood cells.

If mutations in the *G6PD* gene reduce the amount of glucose-6-phosphate dehydrogenase or alter its structure, this enzyme can no longer play its protective role. As a result, reactive oxygen species can accumulate and damage red blood cells.

Factors such as infections, certain drugs, or ingesting fava beans can increase the levels of reactive oxygen species, causing red blood cells to be destroyed faster than the body can replace them. A reduction in the amount of red blood cells causes the signs and symptoms of hemolytic anemia.

Researchers believe that carriers of a *G6PD* mutation may be partially protected against malaria, an infectious disease carried by a certain type of mosquito. A reduction in the amount of functional glucose-6-dehydrogenase appears to make it more difficult for this parasite to invade red blood cells. Glucose-6-phosphate dehydrogenase deficiency occurs most frequently in areas of the world where malaria is common.

Inheritance Pattern

This condition is inherited in an X-linked recessive pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

Other Names for This Condition

- Deficiency of glucose-6-phosphate dehydrogenase
- G6PD Deficiency
- G6PDD
- glucose 6 phosphate dehydrogenase deficiency

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Glucose 6 phosphate dehydrogenase deficiency https://www.ncbi.nlm.nih.gov/gtr/conditions/C0017758/

Other Diagnosis and Management Resources

- Baby's First Test http://www.babysfirsttest.org/newborn-screening/conditions/glucose-6-phosphate-dehydrogenase-deficiency
- MedlinePlus Encyclopedia: Glucose-6-phosphate dehydrogenase deficiency https://medlineplus.gov/ency/article/000528.htm

- MedlinePlus Encyclopedia: Glucose-6-phosphate dehydrogenase test
  https://medlineplus.gov/ency/article/003671.htm
- MedlinePlus Encyclopedia: Hemolytic anemia
  https://medlineplus.gov/ency/article/000571.htm
- MedlinePlus Encyclopedia: Newborn jaundice
  https://medlineplus.gov/ency/article/001559.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Glucose-6-phosphate dehydrogenase deficiency
  https://medlineplus.gov/ency/article/000528.htm
- Encyclopedia: Glucose-6-phosphate dehydrogenase test
  https://medlineplus.gov/ency/article/003671.htm
- Encyclopedia: Hemolytic anemia
  https://medlineplus.gov/ency/article/000571.htm
- Encyclopedia: Newborn jaundice
  https://medlineplus.gov/ency/article/001559.htm
- Health Topic: Anemia
  https://medlineplus.gov/anemia.html
- Health Topic: G6PD Deficiency
  https://medlineplus.gov/g6pddeficiency.html
- Health Topic: Newborn Screening
  https://medlineplus.gov/newbornscreening.html

Genetic and Rare Diseases Information Center

- Glucose-6-phosphate dehydrogenase deficiency
  https://rarediseases.info.nih.gov/diseases/6520/glucose-6-phosphate-dehydrogenase-deficiency

Educational Resources

- Disease InfoSearch: Glucose 6 Phosphate Dehydrogenase Deficiency
  http://www.diseaseinfosearch.org/Glucose+6+Phosphate+Dehydrogenase+Deficiency/3096
- KidsHealth from the Nemours Foundation
  http://kidshealth.org/en/parents/g6pd.html
- MalaCards: hemolytic anemia due to g6pd deficiency
  http://www.malacards.org/card/hemolytic_anemia_due_to_g6pd_deficiency
- Orphanet: Glucose-6-phosphate-dehydrogenase deficiency
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=362

Patient Support and Advocacy Resources

- Children Living With Inherited Metabolic Diseases (CLIMB) (UK)
  http://www.climb.org.uk/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/glucose-6-phosphate-dehydrogenase-deficiency/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/metaboli.html

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22glucose-6-phosphate+dehydrogenase+deficiency%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Glucosephosphate+Dehydrogenase+Deficiency%5BMAJR%5D%29+AND+%28glucose-6-phosphate+dehydrogenase+deficiency%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- GLUCOSE-6-PHOSPHATE DEHYDROGENASE
  http://omim.org/entry/305900

Sources for This Summary

- Beutler E. G6PD deficiency. Blood. 1994 Dec 1;84(11):3613-36. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/7949118
- Dhaliwal G, Cornett PA, Tierney LM Jr. Hemolytic anemia. Am Fam Physician. 2004 Jun 1;69(11): 2599-606. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15202694
- Frank JE. Diagnosis and management of G6PD deficiency. Am Fam Physician. 2005 Oct 1;72(7): 1277-82. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16225031
- Kaplan M, Hammerman C. Glucose-6-phosphate dehydrogenase deficiency: a potential source of severe neonatal hyperbilirubinaemia and kernicterus. Semin Neonatol. 2002 Apr;7(2):121-8. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12208096
- Mehta A, Mason PJ, Vulliamy TJ. Glucose-6-phosphate dehydrogenase deficiency. Baillieres Best Pract Res Clin Haematol. 2000 Mar;13(1):21-38. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10916676
- Ruwende C, Hill A. Glucose-6-phosphate dehydrogenase deficiency and malaria. J Mol Med (Berl). 1998 Jul;76(8):581-8. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9694435
- Verrelli BC, McDonald JH, Argyropoulos G, Destro-Bisol G, Froment A, Drousiotou A, Lefranc G, Helal AN, Loiselet J, Tishkoff SA. Evidence for balancing selection from nucleotide sequence analyses of human G6PD. Am J Hum Genet. 2002 Nov;71(5):1112-28. Epub 2002 Oct 11.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12378426
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC385087/

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